ClinVar

NCBI's ClinVar is a freely available submission-driven database for information about genomic variation and its relationship to human health.

ClinVar accepts submissions interpretations of genetic data from:

- **clinical genetics testing laboratories**
- **research groups**
- **expert panels**
- **and others**

Interpret your data and guide your diagnosis
ncbi.nlm.nih.gov/clinvar

- **1,670+ submitters**
- **75+ countries**
- **841,000+ variants**
- **1,300,000+ submitted records**
- **ClinVar Search Video**

Contact us at clinvar@ncbi.nlm.nih.gov

Follow us on Twitter @ncbi_clinical

Visit us at ncbi.nlm.nih.gov/clinvar/ to find out more

National Library of Medicine
National Center for Biotechnology Information

ClinVar aggregates
clinical assertions about variants provided by clinical genetics testing laboratories
and others.

ClinVar helps clinicians
interpret genetic test results
and diagnose disorders to
improve patient outcomes.

What's New

Tailored notifications for changes in clinical interpretation of variants

Search in ClinVar

Visit Now

- Gene symbols
- HGVS expressions
- RS numbers
- Protein changes
- Diseases and Phenotypes
- Submitting organization
- PubMed ID or other citation

Dowload from the FTP Site

GeneReviews®

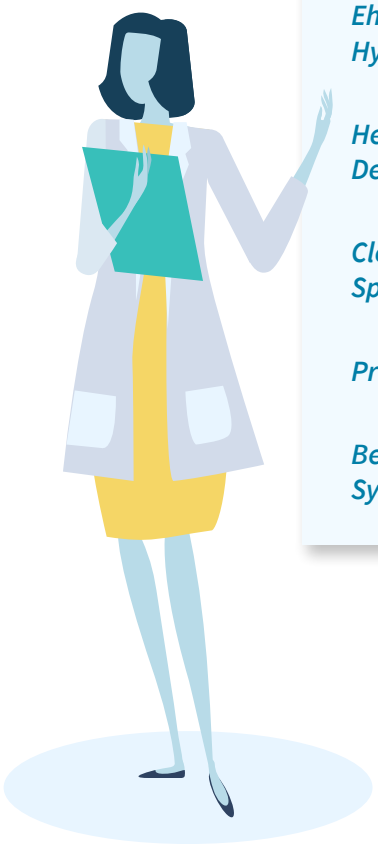
Free, current, expert-authored, and peer-reviewed descriptions of heritable disorders

ABOUT GENEREVIEWS®

- The busy clinician's #1 source for diagnosis, management, and genetic counseling information
- 780+ chapters with links to disease-specific and general consumer resources
- Includes chapters with a single-gene or -phenotype focus and overviews covering the genetic causes of common conditions
- Educational materials to clarify genetics concepts for patients and general clinicians
- Resources for genetics professionals on genetic testing, including founder variants and direct-to-consumer testing
- New and/or updated chapters published weekly
- Content linked to ClinVar, MedGen, GTR®, PheGenI, Medical Genetics Summaries, and dbGaP

Search GeneReviews® By:

- Disorder
- Text
- Author
- Gene Symbol
- Protein Name



GeneReviews® Chapters

Ehlers-Danlos Syndrome, Hypermobility Type

Hereditary Hearing Loss and Deafness Overview

Cleidocranial Dysplasia Spectrum Disorder

Prader-Willi Syndrome

Beckwith-Wiedemann Syndrome

Volunteer to create a GeneReviews® chapter by going to our website and clicking on "For Current/Prospective Authors"

Visit GeneReviews

MedGen

Medical genetics information at your fingertips, aggregated from your favorite resources!

ACCESS RESOURCES

As a student, bookmark MedGen and use it as the starting point to learn about genetic phenotypes and navigate to authoritative resources with information about the phenotype. You can quickly be fully prepared for your patient visits!

SEARCH CLINICAL FEATURES

Search aggregated data by clinical feature(s), genes or other attributes, to help inform a differential diagnosis and hone patient research with access to the latest clinical studies, clinical trials, and systematic reviews

IMPROVE OUTCOMES

Students can quickly access practice guidelines from medical and professional societies that provide effective treatment options for patients as well as consumer resources to educate patient and family about the diagnosis, prognosis, and resources

RESOURCES

MedGen supports research, diagnosis and treatment of genetic disorders by providing information on:

- Mendelian disorders
- Pharmacogenetic responses
- Complex diseases
- Clinical findings

TOOLS

MedGen's all-in-one platform connects clinicians to leading genetic resources, including:

- PubMed
- GARD
- GeneReviews®
- OMIM

Visit MedGen

MedGen, ClinVar, and GTR®

Using NCBI's medical genetics and human variation resources to research, diagnose and treat genetic conditions.

CASE STUDY: SUSPECTED MARFAN SYNDROME

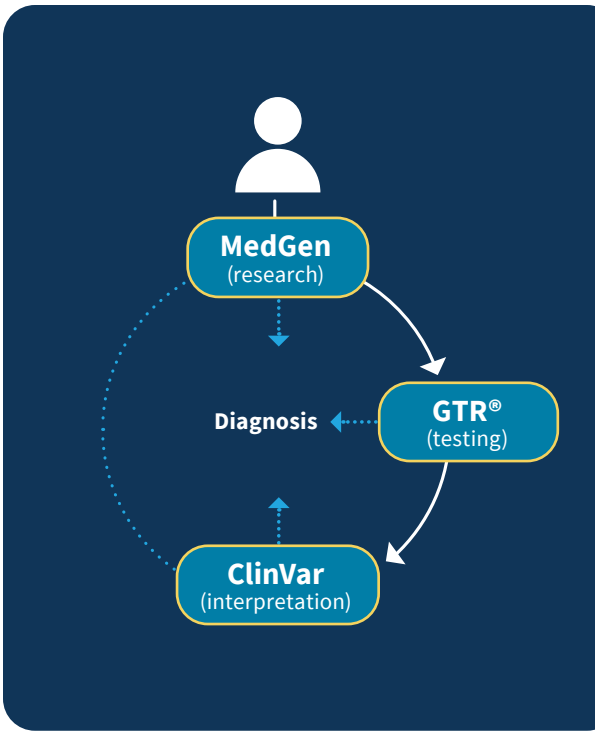
A 9-year-old boy comes into the clinic for a medical release to play soccer. The child presents with myopia, arachnodactyly and has a family history of aortic dissection. These are flags that prompt you to investigate a possible genetic disease.

1 Search MedGen to help develop a differentby patient's clinical features in ial diagnosis and learn about a condition, its diagnosis and etiology

2 Search the NIH Genetic Testing Registry (GTR®) to find the most appropriate genetic test for your patient – for example a panel that includes all the conditions in your differential diagnosis. Learn about the test's validity and utility, and find the ordering information

3 Following testing, compare the test results to interpreted variants in ClinVar and determine pathogenicity

4 Return to MedGen for practice guidelines from medical and professional societies and the latest research to guide your treatment options. Access consumer resources to help your patient and his family understand his diagnosis, prognosis and available help. If the family is interested, check if there are available studies in ClinicalTrials.gov



ABOUT OUR RESOURCES

ClinVar has more than 1,300,000 submitted records representing more than 841,000 unique variants from 1670+ submitters.

GTR® has 76,000+ tests for 16,000+ conditions and 18,500+ genes, from 575+ labs. It includes clinical and research molecular, cytogenetic and biochemical genetic tests.

MedGen helps research thousands of genetic phenotypes including Mendelian disorders, complex diseases, clinical features and drug responses. It aggregates information from authoritative resources so from one website you can access most available clinical, consumer and molecular resources.

Share Your Data with ClinVar

Share Your Data with GTR

Medical Genetics Summaries (MGS)

Free and growing collection of clinical pharmacogenetics summaries. Each summary reviews one drug, it's use and metabolism, the genetic variants that influence how an individual may respond to the drug and aggregates therapeutic guidelines from authoritative sources.

FAST FACTS FOR BUSY CLINICIANS

- Up-to-date, peer-reviewed, actionable pharmacogenetic information
- Access all available therapeutic recommendations based on genotype from the FDA drug label, medical and professional societies, including CPIC, DPWG, ACMG, ASCO
- Introduction gives a clinical context
- Summarizes latest research and evidence for easy consultation at point-of-care
- Synthesizes drug and gene information
- Genetic testing strategies and links to relevant available tests in the NIH Genetic Testing Registry (GTR®)
- Nomenclature table that translates all naming conventions of relevant alleles and links to ClinVar and dbSNP

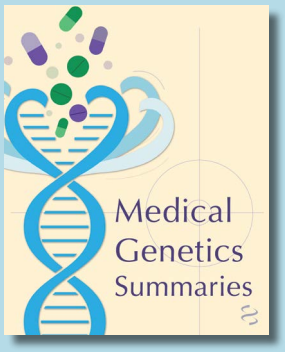
STANDARDIZED FORMAT

- Introduction: Summarizes key points and dosage guidelines
- Gene(s): Genetic variation that influences drug metabolism
- Therapeutic Recommendations based on Genotype excerpted from FDA, CPIC, and other authoritative sources
- Drug: including indications, risks and dosage considerations
- Genetic Testing: Resources for using and interpreting genetic testing
- Nomenclature: Allele translation table including star alleles and HGVS

SAMPLE MGS CHAPTERS

Visit Now

- Codeine
- Tramadol
- Irinotecan
- Tamoxifen
- Pertuzumab
- Flurouracil
- Venlafaxine
- Thioguanine
- Trastuzumab
- Capecitabine
- Mercaptopurine
- **And many, many more…**



MGS VIDEOS:

Pharmacogenetics

Medical Genetics Summaries Overview

Variation Resources

NCBI's variation resources offer human genomic variations, including common and rare SNV, other small-scale variations, large structural variations, and associated frequencies, including ALFA, a new aggregated frequency source based on data from millions of controlled-access research study subjects. Access through the web, APIs, and FTP downloads.

Access data in common bioinformatic standards for teaching courses

Annotate with other data such as genomic features, Genes & Pubmed citations

Open-access data available by web or API

dbSNP

Visit dbSNP

- Over 2 Billion submissions including data from 1000 Genomes, GnomAD, and others
- 720 Million RS
- Frequency for more than 606 Million RS; including common and rare variants
- Rich annotation reported on RefSeq GRCH37 and GRCH38 assemblies, mRNA, and Protein
- VCF files for assemblies GRCh37 and GRCh38
- Full set of RefSNPs in the JSON format
- Indexed Search

dbVar

Visit dbVar

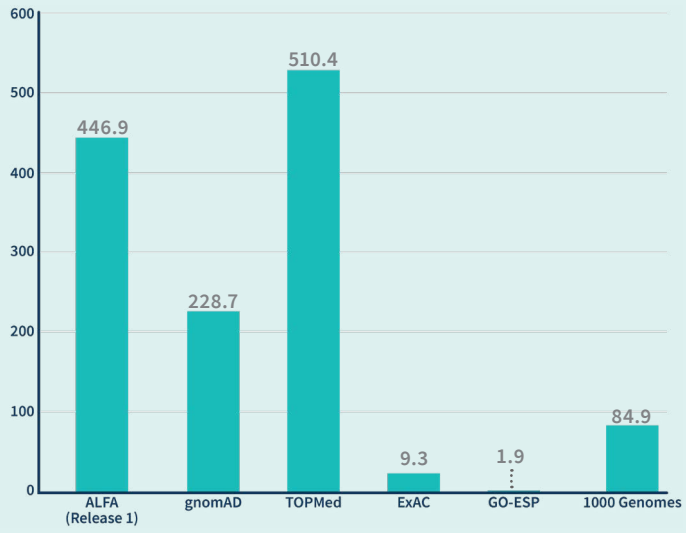
- 193 studies
- Clinically significant SV, Case-Control, and Curated Datasets
- 6.0 million unique structural variants
- 36.1 million submitted variant calls
- Updated monthly
- Population allele frequency
- Files are available in XML, GVF, VCF, BED, BEDPE, and TSV for assemblies GRCh37 and GRCh38
- dbVar Tutorials and Datasets
- Access full set of FTP files

ALFA

Visit ALFA

- Release 1 (March 2020) included 447M variants from 98K subjects
- Release 2 (October 2020) will include an additional ~100K subjects for a total of ~200K
- Access ALFA data along with other projects including GnomAD, and TOPMed

Variants with frequency data (by project in, million)



Variation Services

Web services for comparing, normalizing, annotating, and inter-converting variations

Visit Now

Variation Viewer

View, search, and navigate variations in genomic context. Review data or upload your own data

Visit Now

National Library of Medicine
National Center for Biotechnology Information

Follow us on Twitter @ncbi_clinical

Contact us at info@ncbi.nlm.nih.gov

Visit us at ncbi.nlm.nih.gov/variation/